ABSTRACT OF THE INVENTION

Canavan disease, an autosomal recessive leukodystrophy, is caused by deficiency of aspartoacylase and accumulation of N-acetylaspartic acid in brain. Human aspartoacylase *(ASP)* cDNA spanning 1,435 bp has been cloned and expressed in *E. coli*. A base change, a854>c, has been found in 85% of the 34 Canavan alleles tested so far, which results in a missense glu285>ala mutation that is predicted to be part of the catalytic domain of aspartoacylase. The invention therefore provides nucleic acid sequences, genes, polypeptides, antibodies, vectors containing the gene, host cells transformed with vectors containing the gene, animal models for the disease, methods for expressing the polypeptide, genetic screening methods and kits, diagnostic methods and kits, methods of treating Canavan disease and methods of genetic therapy for the disease.